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The Sword of Damocles: The Psychosocial Impact of Familial Spinocerebellar Ataxia in South Africa

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A survey was conducted on 30 unaffected individuals from a family with autosomal dominant late onset spinocerebellar ataxia in South Africa. The psychological impact of the disorder on individual lives, risk awareness, attitudes towards affected kin and reproduction were evaluated. Respondents employed various psychological strategies to deal with the threat of developing the disorder. In a comparison of "assigned" risk with "perceived" risk, 80% of unaffected persons reported incorrect perceptions of personal risk status. The disorder had little impact on attitudes concerning reproduction; the majority of individuals at risk wanted more children. These issues need to be addressed in the genetic and predictive testing service for familial ataxia in South Africa. Am. J. Med. Genet. 74:270–274, 1997.

KEY WORDS: spinocerebellar ataxia; psychosocial impact; risk awareness; genetic counselling

INTRODUCTION

The familial autosomal dominant late onset spinocerebellar ataxias are a heterogeneous group of progressive neurological disorders in which ataxia is the predominant manifestation. Affected persons suffer from a progressive disturbance of gait, limb ataxia and dysarthria. These symptoms may be accompanied by a variety of clinical manifestations including dementia which may occur late in the course of the illness. The age of onset of the disorder is usually in the third or fourth decade, but may be as early as 16 years [Bryer et al., 1992]. Most affected persons are unable to walk unaided within 15 years of onset, and life expectancy is shortened.

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Late onset autosomal dominant ataxia is not uncommon in the Western Cape region of South Africa. A community nursing sister in a rural area reported a high incidence of presumed anxiety-related complaints among unaffected individuals from several nuclear families in a four-generation kindred with SCA1. This enquiry, which was planned in response to the community sister's request for counselling for family members, was broadened to include other aspects pertinent to genetic counselling, such as risk awareness, attitudes towards affected kin, and reproduction.

MATERIAL AND METHODS

The community nursing sister contacted the families and relayed information concerning mutual expectations. Family members were invited to spend the day at the community clinic. All participants were informed of the objectives of the survey and consent was obtained.

Subjects

Of the 38 people present, 31 at-risk relatives volunteered to be interviewed. Clinical neurological examination was normal in all participants. One subject with pseudoataxia was excluded from the data analysis. Of the remaining subjects, 16 were male and 14 were female. Five subjects had an affected father, 14 had an affected mother, 4 had an affected aunt or uncle, while 4 had an affected grandfather. The ataxia in one father (of 2 subjects) may have been due to alcoholism. Eight people were married, all of whom had children, and 4 were unmarried with children. The mean age of the sample was 24 years (range = 12–35 years) and the mean years of schooling was 9 years (range = 6–12 years). Three subjects had tertiary education. Occupational levels were: professional (N = 2); skilled (N = 2); semi-skilled (N = 10); unskilled (N = 1). Three subjects were unemployed and 12 attended school.

Questionnaire

A questionnaire was designed for the study (Appendix 1). Because of the limited information in the literature on this disorder, questions were structured in an open-ended format (except for those pertaining to psychosomatic complaints) and phrased in such a manner

to allow for free expression. The questionnaire was translated and back-translated into Afrikaans, the mother tongue of most people in this family.

Question 1 investigated the psychosocial impact of the disease on individual family members. Responses were rated by the investigators as: no effect, or uncertain (score = 0); moderate effect (score = 1); major effect (score = 2); or positive effect (score = 3).

Subjects' perceptions of the major problems for the individual (Question 2a) and their families (Question 2b) were assigned to the following descriptive categories: physical disability (difficulty walking, impaired speech, or limb co-ordination); cognitive disability (symptoms of dementia); emotional problems (disorder of mood or behaviour); occupational disability (inability to work and loss of income); social problems (social stigma/embarrassment); burden to the family (having to care for affected family relatives).

The next questions (Questions 3–8) probed risk awareness. Subjects were assigned a risk status (Low = 0, Intermediate = 1, High = 2) by the last author based on the following criteria.

Low risk status: unaffected with an affected grandparent, but an unaffected parent over 49 years of age (N = 7; clinical data in this family indicate a range of age of onset from 16–49 years);

Intermediate Risk Status: unaffected with an affected grandparent and an unaffected parent younger than 49 years of age (N = 4);

High risk status: unaffected with an affected parent (N = 19).

The "perceived" personal risk status (Question 7) responses were categorised and scored as follows: no risk (0); uncertain or possible risk (1); high risk (2).

The "assigned" risk category was determined solely on clinical data from the study of the pedigree (molecular studies had not been completed at the time of the study).

Procedure

The questionnaire was administered to participants individually in their mother-tongue. Subjects younger than 16 years (7 subjects) were not asked about procreation (Question 12).

Responses were subjected to descriptive analysis, and extracts of individual responses were selected to illustrate salient concerns of this family. Particular issues pertinent to prospective genetic counselling were then investigated by means of the Pearson Product-Moment Correlation and the Chi-square and Contingency Coefficient [Siegel, 1956]. The statistical tests were two-tailed and the level of significance was 0.01. These investigations examined: 1) whether psychosocial impact on the individual correlated with age, education, or sex; 2) whether psychosocial impact on the individual was associated with whether they had a first degree (mother, father) or other affected relative (uncle, aunt, or grandfather); 3) whether psychosocial impact correlated with "perceived" risk"; 4) whether "perceived" risk correlated with "assigned risk"; 5) whether there was an association between "perceived" risk and having a first degree or other affected relative.

RESULTS

Psychological Impact of the Disorder on the Lives of Unaffected Family Members

Fifty percent (N = 15) of subjects reported that the disorder had a major negative psychosocial impact on their lives, 16.7% (N = 5) a moderately negative effect, 26.7% (N = 8) were uncertain, or reported no effect. Responses classified as a major negative psychosocial impact related predominantly to fears of the consequences of the disease, unhappiness, and being ashamed or embarrassed about their kin. One person said, "I believe it is a curse". Examples of moderate effects were, "I wonder where the sickness comes from, I have not really paid much attention to it, but sometimes I worry about whether I will get it". Positive effects were elicited in only 6.7% (N = 2). These included a strengthening of religious conviction, and maintaining mutually supportive relationships among family members.

There was no correlation between psychosocial impact and age (r = .236, p = .209), education (r = .359, P = .051) or sex (χ^2 = 2.24, df = 3, C = .263, P = .523). Similarly, there was no association between psychosocial impact and whether the individual had a first degree or other relative (χ^2 = 1.36, df = 3, C = .208, P = .714), or psychosocial impact and "perceived" risk (χ^2 = 4.94, df = 6, C = .376, P = .551).

Eighteen subjects (60%) cited physical disability as the major problems for the affected person, six (20%) a burden to the family, five (17%) cognitive disability, four (13%) emotional problems, four (13%) social problems, and two subjects (6.7%), loss of income. Comments about the patients' inability to care for themselves were grouped under "burden to the family". Cognitive problems were described *inter alia* as, "their minds become smaller and they become like a child", or "does not think right, talks mixed up". In one family, the affected mother was emotionally labile, short-tempered and prone to outbursts of temper. Social problems reported across families included being laughed at, feeling socially isolated in the family, or being falsely accused of being inebriated in public.

Major problems for the whole family were emotional problems (N = 11, 36.7%), burden to the family (N = 11, 36.7%), and occupational limitations (N = 2, 6.7%). The emotional problems mentioned varied. The collective fear was voiced by one person who said, "Everyone thinks, will I, or will I not get it". Two respondents worried about early death ("the problem is that they all die"), while the same family who reported emotional problems in their mother, were burdened by having to cope with her emotional lability.

Risk Awareness

Ten (33.3%) subjects believed that no other family members would develop the disorder, 18 (60%) that other family members would, and 2 (6.7%) were uncertain. Five subjects assigned the risk to a parent, 11 to a sibling, 2 to an aunt or uncle. The assignments of risk to a particular family member were based on supposed

emotional and physical characteristics shared with an affected relative (for example: "she has a similar nature"; "he walks the same way"; "she has headaches"; "he stutters"; "she has a bewildered look"; or, "she has a nervous disposition").

All participants correctly reported that they did not have the disorder. Eighteen respondents (60%) unequivocally stated they would not get the disorder. Eleven subjects (36.7%) were uncertain or foresaw a possible risk, while one subject (3.3%) believed he would develop the disorder. Some respondents submitted more than one reason for believing they were not at risk. Reasons included: religious beliefs (N = 5); regular participation in sport (N = 4); that he/she was healthy and was free of symptoms (N = 5); that a parent had said he would not get the disease (N = 1). Three respondents gave non-specific answers, and two did not respond to this question.

Of the 12 individuals who perceived a possible or positive risk for developing the disease, only three cited genetic reasons (viz. they knew the disease was inherited, but were unsure of the absolute risk). Two persons thought they may be at risk because of physical problems such as pains and "lameness" in their legs and knees. One person believed that by taking calcium powders to strengthen her limbs, she may ward off the disorder. Five respondents were uncertain why they were at risk. The person who believed he would develop the disease, said his father had told him so. There was no correlation between "perceived" risk and whether they had a first degree or other affected relative (χ^2 = 3.27. df = 2, C = .313, P = .195).

Similarly, "perceived" risk did not correlate with "assigned" risk (χ^2 = 2.76, df = 4, C = .29, P = .598). Twenty-three of the 30 subjects had incorrect perceptions about their risk status. Notably, of the 19 (63.3%) subjects who were assigned a high risk status, 13 (43.33%) saw themselves as not at risk, and five (16.67%) at possible risk. One subject correctly said he was at high risk. Four (13.33%) subjects with a low risk status saw themselves at possible risk.

Attitudes Towards Affected Family Members

Most respondents felt compassion and concern for the affected relative. Three people who had fears for the future of their affected relatives, added that in observing them, they were continually reminded of the potential threat to their own lives. One person felt duty-bound to care for affected relatives, while another confessed to difficulty coping with them. Four respondents admitted they were embarrassed by their affected relatives. Another respondent worried that her affected mother was unable to fulfil maternal duties.

More than half (N = 17) of the subjects reported feeling "happy" or "glad" when they considered unaffected family members. In a few respondents, this was tempered by insecurity about the future, for example: "those who have the illness were well at first"; "you never know what will happen to them"; "it would be bad if everyone gets it". A few persons distanced themselves from affected relatives ("I keep myself apart", or "I'm glad I am healthy"). Two respondents felt that those who were spared had a responsibility to look after affected relatives. One subject was ashamed and angry at those who were healthy but did not work, because, "my mother, who is sick, still works". Another worried about the future of the children of affected parents.

Psychosomatic Complaints

Seventy percent of unaffected persons reported at least one psychosomatic complaint (Table I). The most commonly reported symptom was nervousness, followed by headaches and moodiness.

Attitudes Towards Reproduction

The majority of unaffected persons (69.6%) wanted more children (Table II), 11 of whom planned on having two or more. Only six of these 16 respondents referred to the familial illness. Three subjects said they wanted more children, but not if they developed the illness, one of whom said she worried that her children would reproach her. Two others said they planned to have children whether they developed the disorder or not. The sixth person said he did not consider the family disease when he had decided to have children.

Comparing risk awareness and attitudes towards procreation in these six people, of the three who said they would not have children if they developed the disorder, two had earlier rated themselves as at "no risk", and the other one, as at "uncertain/possible risk". The person who said he had not thought of the familial disease also perceived himself to be at "possible risk". The two persons who said they would have children even if they got the disorder, saw themselves as at "no risk".

All six unaffected people who did not want more children already had children. The risk for the developing the disease was cited by two of these six subjects. Other reasons given were financial, physical (for example, sterilization, or other medical complaints), and a completed family.

DISCUSSION

We report our findings on a large family from a low socio-economic background with low levels of education who were interviewed by questionnaire on the psychosocial aspects of late onset familial spinocerebellar ataxia. In compiling the questionnaire, we avoided the use of scientific terminology since we did not wish to burden respondents with phrases foreign to them. (For the purposes of this paper, responses had to be trans-

TABLE I. Psychosomatic Complaints in Unaffected Persons

Symptoms	N	Percentage
Headache	11	36.7
Stomach ache	3	10.0
Tearfulness	8	26.7
Sleep disturbance	2	6.7
Nervousness	12	40.0
Mood swings	11	36.7

TABLE II. Attitudes of Unaffected Individuals Towards Reproduction

Response	N	Percentage
Want more children	16	69.6
No further children	6	26.1
Uncertain	1	4.4
TOTAL	23	

lated from Afrikaans into English, and, as always, the full impact of quotes is lost in the translation process). The response to our study was favourable; 81.6% of people attending the clinic volunteered to be interviewed.

Contrary to earlier studies on kindred with familial ataxia (SCA1) who overestimated their genetic risk [Nance et al., 1994], the majority of our respondents were unaware or denied the reality of their personal risk status. There were various reasons why this occurred. This family lives in a rural area and access to specialised services is limited, not only due to the distance, but also because of their financial constraints. As far as we were aware, unaffected relatives had received no formal genetic counselling and were dependent on affected relatives who had been seen in the tertiary care service for information on the disorder. Also, there was a complex interplay between being ill-informed and various psychological strategies used by people at risk. These strategies included defence mechanisms as described in the literature in Huntington disease [Wexler, 1992; Kesler, 1988; Martindale, 1987].

Defence mechanisms are frequently seen as maladaptive in clinical practice and in the medical literature, despite evidence that they can be healthy coping mechanisms promoting personal health in medical crises [Russell, 1993; Greer, 1992; Fricchione et al., 1992]. For people living with the threat of spinocerebellar ataxia, the absence of defence mechanisms could render them helpless in an impossible situation. Denial not only allays anxiety, but also allows for hope and enthusiasm. Despite the threat of developing the disorder, most at risk people were either employed or students. There were many indications that family members refused to be passive victims and took active steps to control their lives. They participated regularly in sport, used ointment, or ingested calcium to strengthen their legs. Others turned to religion, trusting that God would spare them the illness. Turning to "a higher order for control" to contain fear and anxiety has been previously described in people at risk for Huntington disease [Wexler, 1979].

In this context of victim versus active agent in controlling one's fate, the fact that no respondents had chosen not to have children because of the disease is of interest. Some subjects said they wanted children even though they might develop the family illness. Previous authors have noted that knowledge of the risk of Huntington disease does not necessarily alter the attitude of those who plan to have a family [Barette and Marsden, 1979], some of whom have expressed their incomprehension at the irrationality of continued reproduction despite the illness [Pearson, 1973]. Our impression was that in this family, having children bestowed people with a sense of normality and empowerment in an otherwise abnormal situation.

In order to secure the belief that their lives could be normal, and that they would not develop the disorder, more than a third of this cohort projected their fears of developing this disease onto other family members. As noted in Machado-Joseph disease (a similar disorder with a different phenotype), resemblance to an affected relative's physique and/or temperament, was not infrequently a measure of perceived risk [Boutté, 1987]. Because the family illness is associated with brain disease, having headaches was also seen as an ominous sign.

Most persons expressed positive sentiments towards their affected relatives, but not unexpectedly, a few persons avoided contact with them. Feelings of resentment, suspicion and embarrassment were occasionally elicited towards healthy members of the family. One person told us that her grandmother had wondered whether those who were healthy were concealing preventative measures they were ingesting. In this context, the person with pseudoataxia who was excluded from the study, is of interest. His "symptoms" (which were not as disabling as that of his siblings) afforded him the status of the affected, and protected him from the possibility of being rejected as a survivor. For him, living in a family with this hereditary disorder was a double-edged sword.

In addition to the insights gained into the psychological strategies employed, we learnt that it cannot be simply assumed that western-type genetic counselling is appropriate in Africa. All too frequently, this proceeds in a medical model using medical terminology that is alien to lay people. The study familiarised us with the language that this family used to describe their concerns and on our return to the clinic, we conveyed the emotive concept of genetic risk in a discourse that was accessible to them. Since this study, molecular studies have provided an accurate means for presymptomatic and prenatal testing for the disorder [Orr et al., 1993; Ranum et al., 1994]. A predictive service for both Huntington and spinocerebellar ataxia is now available at this hospital and certain members of this family have elected to enter the program.

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Appendix 1: Questionnaire

1. How did the family illness influence your life?
2. What do you think is the biggest problem for:
   a) the person with the illness?
   b) the family?
3. Who in the family do you think will get it?
4. Why do you think so?
5. Did anybody ever say that you will get it?
6. Who, and why did they say so?
7. Do you think that you will get it, or that you already have it?
8. Why do you think so?
9. How do you feel if you think of, or look at your mother/father/brother/sister, or any other family member who has it?
10. And about those who are healthy?
11. Have you ever had any of the following problems? How often?
    a) Headaches
    b) Stomach aches
    c) Tearfulness
    d) Difficulty sleeping
    e) Nervousness
    f) Moodiness
    g) Any other problems
12. Do you want more children?
    a) If yes, how many?
    b) If no, why not?